



## 2q37 deletion syndrome

2q37 deletion syndrome is a condition that can affect many parts of the body. This condition is characterized by weak muscle tone (hypotonia) in infancy, mild to severe intellectual disability and developmental delay, behavioral problems, characteristic facial features, and other physical abnormalities.

Most babies with 2q37 deletion syndrome are born with hypotonia, which usually improves with age. About 25 percent of people with this condition have autism, a developmental condition that affects communication and social interaction.

The characteristic facial features associated with 2q37 deletion syndrome include a prominent forehead, highly arched eyebrows, deep-set eyes, a flat nasal bridge, a thin upper lip, and minor ear abnormalities. Other features of this condition can include short stature, obesity, unusually short fingers and toes (brachymetaphalangy), sparse hair, heart defects, seizures, and an inflammatory skin disorder called eczema. A few people with 2q37 deletion syndrome have a rare form of kidney cancer called Wilms tumor. Some affected individuals have malformations of the brain, gastrointestinal system, kidneys, or genitalia.

### Frequency

2q37 deletion syndrome appears to be a rare condition, although its exact prevalence is unknown. Approximately 100 cases have been reported worldwide.

### Genetic Changes

2q37 deletion syndrome is caused by a deletion of genetic material from a specific region in the long (q) arm of chromosome 2. The deletion occurs near the end of the chromosome at a location designated 2q37. The size of the deletion varies among affected individuals. The signs and symptoms of this disorder are probably related to the loss of multiple genes in this region.

### Inheritance Pattern

Most cases of 2q37 deletion syndrome are not inherited. They result from a chromosomal deletion that occurs as a random event during the formation of reproductive cells (eggs or sperm) or in early fetal development. Affected people typically have no history of the disorder in their family.

Rarely, affected individuals inherit a copy of chromosome 2 with a deleted segment from an unaffected parent. In these cases, one of the parents carries a chromosomal rearrangement between chromosome 2 and another chromosome. This rearrangement

is called a balanced translocation. No genetic material is gained or lost in a balanced translocation, so these chromosomal changes usually do not cause any health problems. However, translocations can become unbalanced as they are passed to the next generation. Children who inherit an unbalanced translocation can have a chromosomal rearrangement with extra or missing genetic material. Some individuals with 2q37 deletion syndrome inherit an unbalanced translocation that deletes genetic material near the end of the long arm of chromosome 2, which results in birth defects and other health problems characteristic of this disorder.

### **Other Names for This Condition**

- Albright hereditary osteodystrophy-like syndrome
- brachydactyly-mental retardation syndrome

### **Diagnosis & Management**

These resources address the diagnosis or management of 2q37 deletion syndrome:

- GeneReview: 2q37 Microdeletion Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1158>
- Genetic Testing Registry: Brachydactyly-Mental Retardation syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838126/>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>

## **Additional Information & Resources**

### MedlinePlus

- Health Topic: Autism Spectrum Disorder  
<https://medlineplus.gov/autismspectrumdisorder.html>
- Health Topic: Developmental Disabilities  
<https://medlineplus.gov/developmentaldisabilities.html>
- Health Topic: Eczema  
<https://medlineplus.gov/eczema.html>
- Health Topic: Wilms Tumor  
<https://medlineplus.gov/wilmstumor.html>

### Genetic and Rare Diseases Information Center

- 2q37 deletion syndrome  
<https://rarediseases.info.nih.gov/diseases/10202/2q37-deletion-syndrome>

### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Seizures and Epilepsy: Hope Through Research  
<https://www.ninds.nih.gov/Disorders/Patient-Caregiver-Education/Hope-Through-Research/Epilepsies-and-Seizures-Hope-Through>

### Educational Resources

- Boston Children's Hospital: Autism  
<http://www.childrenshospital.org/conditions-and-treatments/conditions/autism-spectrum-disorders>
- Disease InfoSearch: Brachydactyly-Mental Retardation syndrome  
<http://www.diseaseinfosearch.org/Brachydactyly-Mental+Retardation+syndrome/1525>
- MalaCards: 2q37 deletion syndrome  
[http://www.malacards.org/card/2q37\\_deletion\\_syndrome](http://www.malacards.org/card/2q37_deletion_syndrome)
- March of Dimes: Chromosomal Conditions  
<http://www.marchofdimes.org/baby/chromosomal-conditions.aspx>
- Orphanet: 2q37 microdeletion syndrome  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=1001](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1001)
- Unique support pamphlet about 2q37 deletion syndrome  
<http://www.rarechromo.org/information/Chromosome%20%202/2q37%20deletion%20syndrome%20FTNW.pdf>

## Patient Support and Advocacy Resources

- Autism Society of America  
<http://www.autism-society.org/>
- Chromosome Disorder Outreach  
<http://chromodisorder.org/>
- Magic Foundation  
<https://www.magicfoundation.org/>
- Unique: Rare Chromosome Disorder Support Group (UK)  
<http://www.rarechromo.org/html/home.asp>

## GeneReviews

- 2q37 Microdeletion Syndrome  
<https://www.ncbi.nlm.nih.gov/books/NBK1158>

## Genetic Testing Registry

- Brachydactyly-Mental Retardation syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1838126/>

## Scientific articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%282q37%5BTI%5D%29+AND+%28deletion%5BTI%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

## OMIM

- CHROMOSOME 2q37 DELETION SYNDROME  
<http://omim.org/entry/600430>

## **Sources for This Summary**

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